XII CONFERENCIA ANUAL
DE LAS PLATAFORMAS TECNOLÓGICAS
DE INVESTIGACIÓN BIOMÉDICA

Barcelona Supercomputing Center
Centro Nacional de Supercomputación

EXCELENCIA SEVERO OCHOA

Alfonso Valencia. Ph.D.

ICREA Professor

Director Life Sciences Dept.

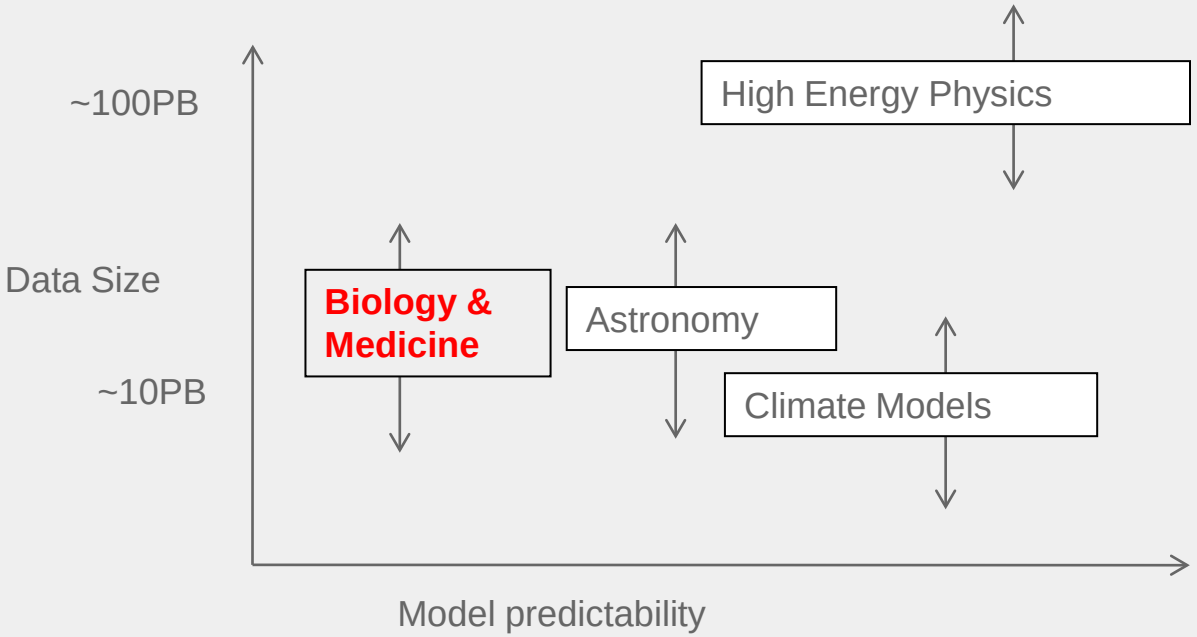
Director Spanish Bioinformatics Institute INB-ISCIII ELIXIR-ES

Big data en investigación biomedica y en práctica clínica.

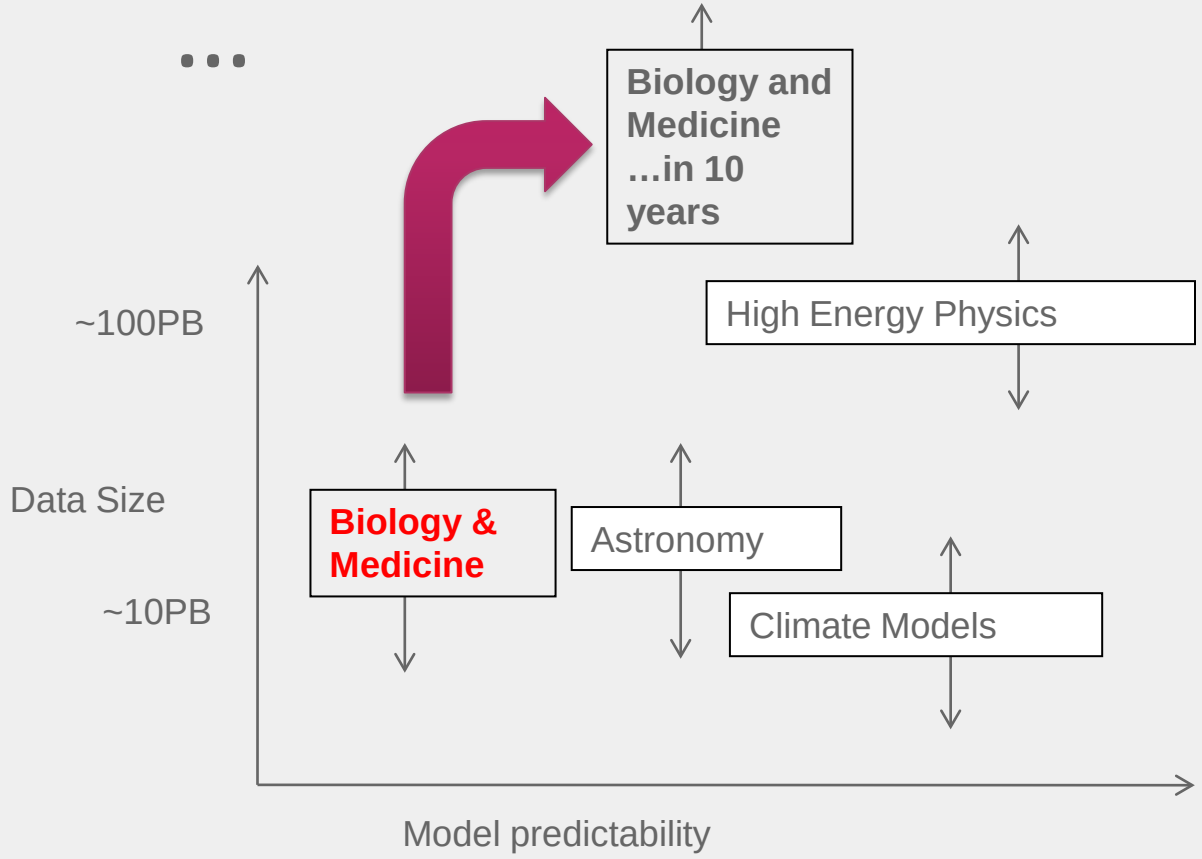
5/March/2019

XII Conferencia Anual de las Plataformas Tecnol6gicas de Investigaci6n Biomedica: Medicamentos Innovadores, Nanomedicina Tecnologfa Sanitaria y Mercados Biotecnol6gicos

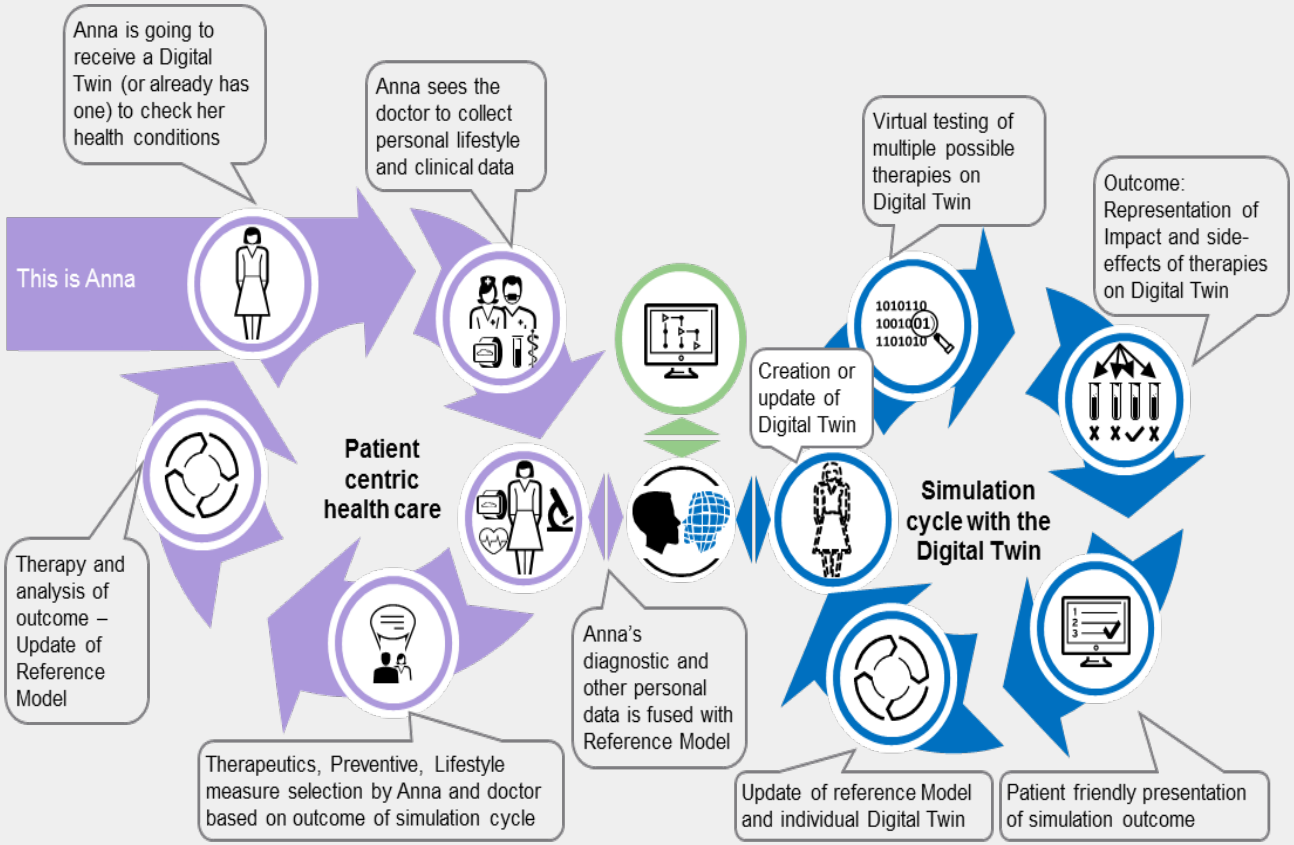
Biology and Medicine as Big Data science



From Ewan Birney EBI


...
Biology and Medicine …in 10 years
High Energy Physics
~100PB
Data Size
~10PB
Biology & Medicine
Astronomy
Climate Models
Model predictability

Digital Twins view of Future Medicine



Screenshot

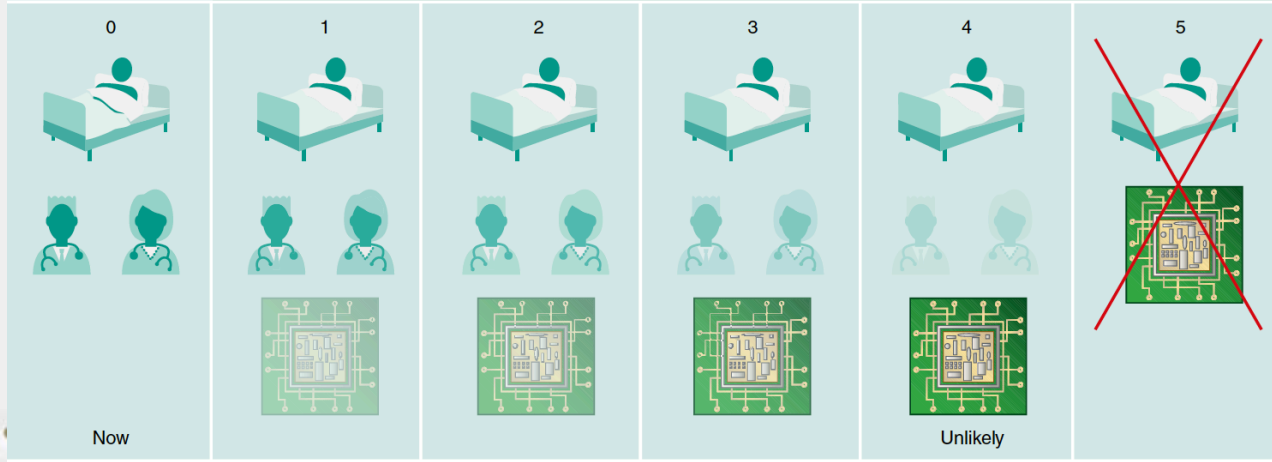
High-performance medicine: the convergence of human and artificial intelligence

NATURE MEDICINE | VOL 25 | JANUARY 2019 | 44–56 |

Eric J. Topol Department of Molecular Medicine, Scripps Research, La Jolla,

Human driver monitors environment			System monitors environment		
0	1	2	3	4	5
No automation	Driver assistance	Partial automation	Conditional automation	High automation	Full automation
The absence of any assistive features such as adaptive cruise control.	Systems that help drivers maintain speed or stay in lane but leave the driver in control.	The combination of automatic speed and steering control—for example, cruise control and lane keeping.	Automated systems that drive and monitor the environment but rely on a human driver for backup.	Automated systems that do everything—no human backup required—but only in limited circumstances.	The true electronic chauffeur: retains full vehicle control, needs no human backup, and drives in all conditions.

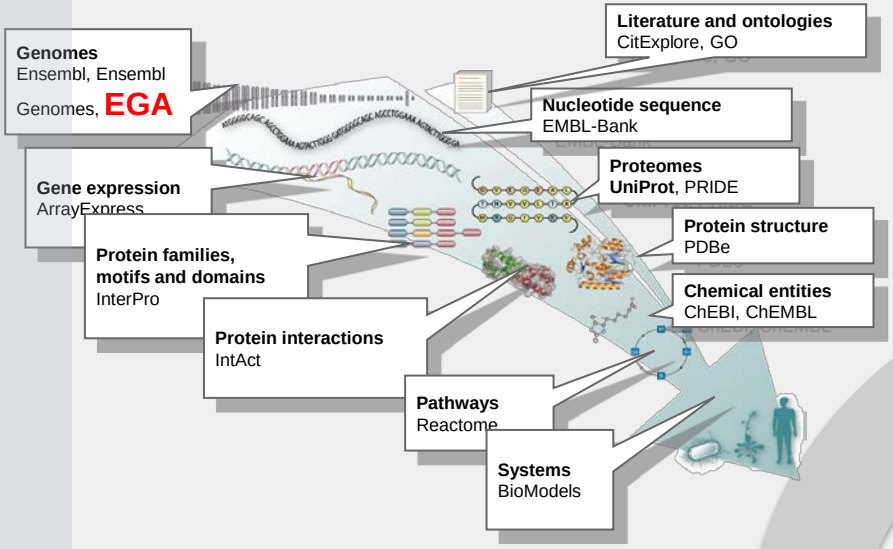
Humans and machine doctors

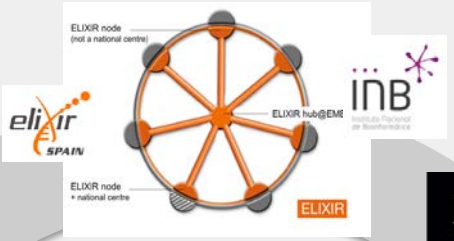


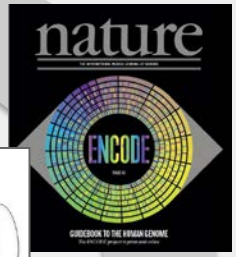
.. Datos muy especiales

- Complejidad asociada a experimentos y muestras (meta-información)
- Heterogeneidad propia de la biología
- Confidencialidad
- Propiedad y gestión de los datos

Large Scale Genomics

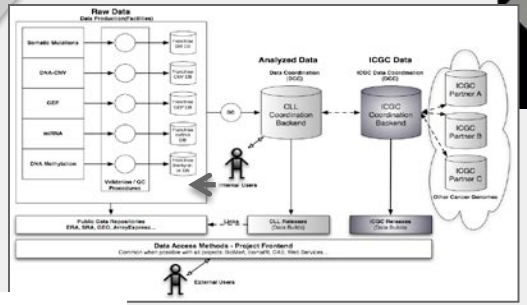






IHEC

BLUEPRINT epigenome



BSC CNAG EBI-EMBL

RD Connect

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

International network of cancer genome projects

The International Cancer Genome Consortium*

ARGO

XII CONFERENCIA ANUAL DE LAS PLATAFORMAS TECNOLÓGICAS DE INVESTIGACIÓN BIOMÉDICA

Barcelona Supercomputing Center
Centro Nacional de Supercomputación

CLL
CHRONIC LYMPHOCYTIC LEUKEMIA
GENOME PROJECT

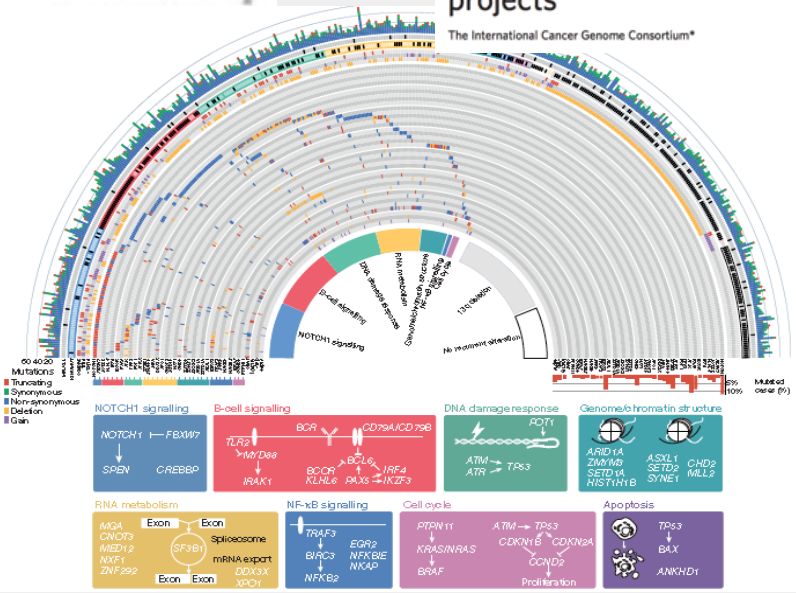
Vol 464|15 April 2010|doi:10.1038/nature08987 nature

International Cancer Genome Consortium

PERSPECTIVES

International network of cancer genome projects

The International Cancer Genome Consortium*



nature
International weekly journal of science

Non-coding recurrent mutations in chronic lymphocytic leukaemia

Xose S. Puente[1], Silvia Beà[2], Rafael Valdés Mas[1], Neus Villamor[3], Jesús Gutiérrez Abril[1], José I. Martín Subero[4], Marta Munar[5], Carlota Rubio Pérez[6], Pedro Jares[7], Marta Aymerich[3], Tycho Baumann[8], Renée Beekman[2], Laura Belver[9], Anna Carrio[2], Giancarlo Castellano[2], Guillem Clot[2], Enrique Colado[10], Dolors Colomer[3], Dolors Costa[2], Julio Delgado[6], Anna Enjuanes[2], Xavier Estivill[11], Adolfo A. Ferrando[9], Josep L. Gelpí[6], Blanca González[2], Santiago González[6], Marcos González[12], Marta Gut[13], Jesús M. Hernández Rivas[12], Mónica López Guerra[3], David Martín García[1], Alba Navarro[1], Pilar Nicolás[14], Modesto Orozco[5], Ángel R. Payer[10], Magda Pinyol[3], David G. Pisano[15], Diana A. Puente[3], Ana C. Queirós[4], Víctor Quesada[1], Carlos M. Romeo Casabona[14], Cristina Royo[2], Romina Royo[2], María Rozman[3], María Russiñol[6], Itziar Salaverría[2], Kostas Stamatopoulos[16], Hendrik G. Stunnenberg[17], David Tamborero[6], María J. Terol[18], Alfonso Valencia[15], Núria López Bigas[6], David Torrents[15], Ivo Gut[13], Armando López Guillermo[3], Carlos López Otín[1]§ & Elías Campo[3]§

nature
International weekly journal of science

nature
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Archive > Volume 475 > Issue 7354 > Letters > Article
NATURE | LETTER OPEN
previous article next article

Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia

Xose S. Puente, Magda Pinyol, Víctor Quesada, Laura Conde, Gonzalo R. Ordóñez, Neus Villamor, Georgia Escaramis, Pedro Jares, Silvia Beà, Marcos González-Díaz, Laia Bassaganyas, Tycho Baumann, Manel Juan, Mónica López-Guerra, Dolors Colomer, José M. C. Tubío, Cristina López, Alba Navarro, Cristian Tornador, Marta Aymerich, María Rozman, Jesús M. Hernández, Diana A. Puente, José M. P. Freije, Gloria Velasco, Ana Gutiérrez-Fernández, Dolors Costa, Anna Carrió, Sara Guijarro, Anna Enjuanes, Lluis Hernández, Jordi Yagüe, Pilar Nicolás, Carlos M. Romeo-Casabona, Heinz Himmelbauer, Ester Castillo, Juliana C. Dohm, Silvia de Sanjosé, Miguel A. Piris, Enrique de Alava, Jesús San Miguel, Romina Royo, Josep L. Gelpí, David Torrents, Modesto Orozco, David G. Pisano, Alfonso Valencia, Roderic Guigó, Mónica Bayés, Simon Heath, Marta Gut, Peter Klatt, John Marshall, Keiran Raine, Lucy A. Stebbings, P. Andrew Futreal, Michael R. Stratton, Peter J. Campbell, Ivo Gut, Armando López-Guillermo, Xavier Estivill, Emili Montserrat, Carlos López-Otín & Elías Campo > Show fewer authors
Affiliations | Contributions | Corresponding authors

Nature 475, 101–105 (07 July 2011) | doi:10.1038/nature10113
Received 16 November 2010 | Accepted 06 April 2011 | Published online 05 June 2011

LETTERS
nature genetics
VOLUME 44 | NUMBER 11 | NOVEMBER 2012 NATURE GENETICS

Epigenomic analysis detects widespread gene-body DNA hypomethylation in chronic lymphocytic leukemia

Marta Kulis[1,11], Simon Heath[2,11], Marina Bibikova[3,11], Ana C Queirós[4,11], Alba Navarro[1], Guillem Clot[1], Alejandra Martínez-Trillos[1], Giancarlo Castellano[1], Isabelle Brun-Heath[5], Magda Pinyol[6], Sergio Barberán-Soler[7], Panagiotis Papasissos[8], Pedro Jares[1], Silvia Beà[1], Daniel Rico[9], Simone Ecker[9], Miriam Rubio[9], Romina Royo[9], Vincent Ho[3], Brandy Klotzle[3], Lluis Hernández[1], Laura Conde[1], Mónica López-Guerra[1], Dolors Colomer[1], Neus Villamor[1], Marta Aymerich[1], María Rozman[1], Mónica Bayés[2], Marta Gut[2], Josep L Gelpí[9], Modesto Orozco[9], Ian-Jing Fan[3], Víctor Quesada[10], Xose S Puente[10], David G Pisano[9], Alfonso Valencia[9], Armando López-Guillermo[1], Ivo Gut[2], Carlos López-Otín[10], Elías Campo[1] & José I Martín-Subero[4]

nature genetics
nature.com > journal home > archive > issue > letter > full text
NATURE GENETICS | LETTER

Exome sequencing identifies recurrent mutations of the splicing factor *SF3B1* gene in chronic lymphocytic leukemia

Víctor Quesada, Laura Conde, Neus Villamor, Gonzalo R Ordóñez, Pedro Jares, Laia Bassaganyas, Andrew J Ramsay, Silvia Beà, Magda Pinyol, Alejandra Martínez-Trillos, Mónica López-Guerra, Dolors Colomer, Alba Navarro, Tycho Baumann, Marta Aymerich, María Rozman, Julio Delgado, Eva Giné, Jesús M Hernández, Marcos González-Díaz, Diana A Puente, Gloria Velasco, José M P Freije, José M C Tubío, Romina Royo, Josep L Gelpí, Modesto Orozco, David G Pisano, Jorge Zamora, Miguel Vázquez, Alfonso Valencia, Heinz Himmelbauer, Mónica Bayés, Simon Heath, Marta Gut, Ivo Gut, Xavier Estivill, Armando López-Guillermo, Xose S Puente, Elías Campo & Carlos López-Otín
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Affiliations | Contributions | Corresponding authors

Nature Genetics 44, 47–52 (2012) | doi:10.1038/ng.1032
Received 12 July 2011 | Accepted 10 November 2011 | Published online 11 December 2011

CSH PRESS GENOME RESEARCH

Transcriptome characterization by RNA sequencing identifies a major molecular and clinical subdivision in chronic lymphocytic leukemia

Pedro G. Ferreira, Pedro Jares, Daniel Rico, et al.

Genome Res. published online November 21, 2013
Access the most recent version at doi:10.1101/gr.152132.112

Eur J Haematol. 2016 Aug;97(2):175-82. doi: 10.1111/ejh.12700. Epub 2015 Dec 17.

LPL gene expression is associated with poor prognosis in CLL and closely related to NOTCH1 mutations.

Kristensen L[1], Kristensen T[1], Abildgaard N[2], Royo C[3], Frederiksen M[4], Mourits-Andersen T[5], Campo E[3], Møller MB[1].

The splicing modulator sudemycin induces a specific antitumor response and cooperates with ibrutinib in chronic lymphocytic leukemia.

Xargay-Torrent S[1], López-Guerra M[1,2], Rosich L[1], Montraveta A[1], Roldán J[1], Rodríguez V[1], Villamor N[2], Aymerich M[2], Lagisetti C[3], Webb TR[3], López-Otín C[4], Campo E[2], Colomer D[1,2].

Leukemia. 2015 Jan;29(1):96-106. doi: 10.1038/leu.2014.143. Epub 2014 Apr 30.

The γ-secretase inhibitor PF-03084014 combined with fludarabine antagonizes migration, invasion and angiogenesis in NOTCH1-mutated CLL cells.

López-Guerra M[1], Xargay-Torrent S[1], Rosich L[1], Montraveta A[1], Roldán J[1], Matas-Céspedes A[1], Villamor N[2], Aymerich M[2], López-Otín C[3], Pérez-Galán P[1], Roué G[1], Campo E[2], Colomer D[4].

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Nadeu F[1], Delgado J[2], Royo C[1], Baumann T[3], Stankovic T[4], Pinyol M[5], Jares P[2], Navarro A[1], Martín-García D[1], Beà S[1], Salaverria I[1], Oldreive C[4], Aymerich M[2], Suárez-Cisneros H[5], Rozman M[2], Villamor N[2], Colomer D[2], López-Guillermo A[2], González M[6], Alcoceba M[6], Terol MJ[7], Colado E[8], Puente XS[9], López-Otín C[9], Enjuanes A[5], Campo E[10].

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López C[1], Delgado J, Costa D, Villamor N, Navarro A, Cazorla M, Gómez C, Arias A, Muñoz C, Cabezas S, Baumann T, Rozman M, Aymerich M, Colomer D, Pereira A, Cobo F, López-Guillermo A, Campo E, Carrió A.

Blood. 2016 Mar 24;127(12):1611-3. doi: 10.1182/blood-2015-10-678490. Epub 2016 Feb 4.

Clinical impact of MYD88 mutations in chronic lymphocytic leukemia.

Martínez-Trillos A[1], Navarro A[2], Aymerich M[3], Delgado J[3], López-Guillermo A[3], Campo E[3], Villamor N[3].

Author information

Blood. 2014 Jun 12;123(24):3790-6. doi: 10.1182/blood-2013-12-543306. Epub 2014 Apr 29.

Mutations in TLR/MYD88 pathway identify a subset of young chronic lymphocytic leukemia patients with favorable outcome.

Martínez-Trillos A[1], Pinyol M[2], Navarro A[2], Aymerich M[1], Jares P[2], Juan M[3], Rozman M[1], Colomer D[1], Delgado J[4], Giné E[4], González-Díaz M[5], Hernández-Rivas JM[5], Colado E[6], Rayón C[6], Payer AR[6], Terol MJ[7], Navarro B[7], Quesada V[8], Puente XS[8], Rozman C[9], López-Otín C[8], Campo E[10], López-Guillermo A[4], Villamor N[1].

Blood. 2015 Jul 9;126(2):195-202. doi: 10.1182/blood-2014-10-604959. Epub 2015 Jun 1.

Mutations in CHD2 cause defective association with active chromatin in chronic lymphocytic leukemia.

Rodríguez D[1], Bretones G[1], Quesada V[1], Villamor N[2], Arango JR[1], López-Guillermo A[2], Ramsay AJ[1], Baumann T[2], Quirós PM[1], Navarro A[2], Royo C[2], Martín-Subero JI[2], Campo E[3], López-Otín C[1].

Personalized Medicine schema

RNAseq
Epigenomics
HiC
Arrays
Exomes
Complete genomes
Metagenomes
Single Cells
Computational Infrastructure
Biomedical repositories of Data and Methods
Bioinformatics Infrastructure
Computational Methods (ML, Cognitive Computing)
Research Interest
Practical utility in the clinic
Mining EHR
Social Media
Medical Imaging
Other Health Data (papers, patents, etc)
Medical devices
Epidemiology

Gomez-Lopez (2017) Briefings in Bioinformatics,

ELIXIR:
European Bioinformatics Infrastructure

- *Data*
- *Interoperability*
- *Tools*
- *Compute*
- *Training*

- *Marine metagenomics*
- *Crop and forest plants*
- ***Human data***
- ***Rare diseases***

ELIXIR Human genomics platform

Generate
Diverse data from diverse providers

Archive, discover, manage
Store, access and share data of multiple types and origin

Analyse

EUROPEAN GENOME-PHENOME ARCHIVE

LOCAL EGA

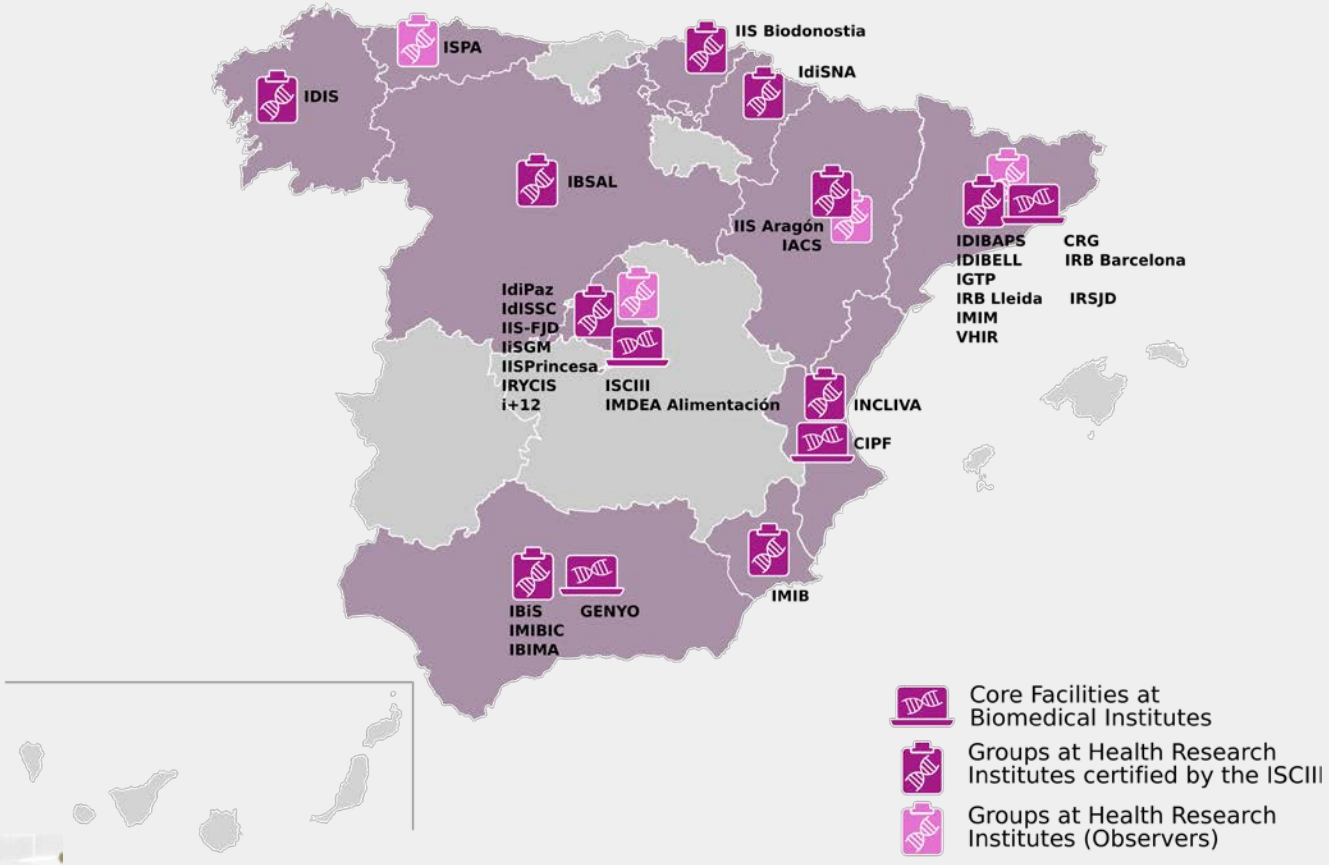
Beacon

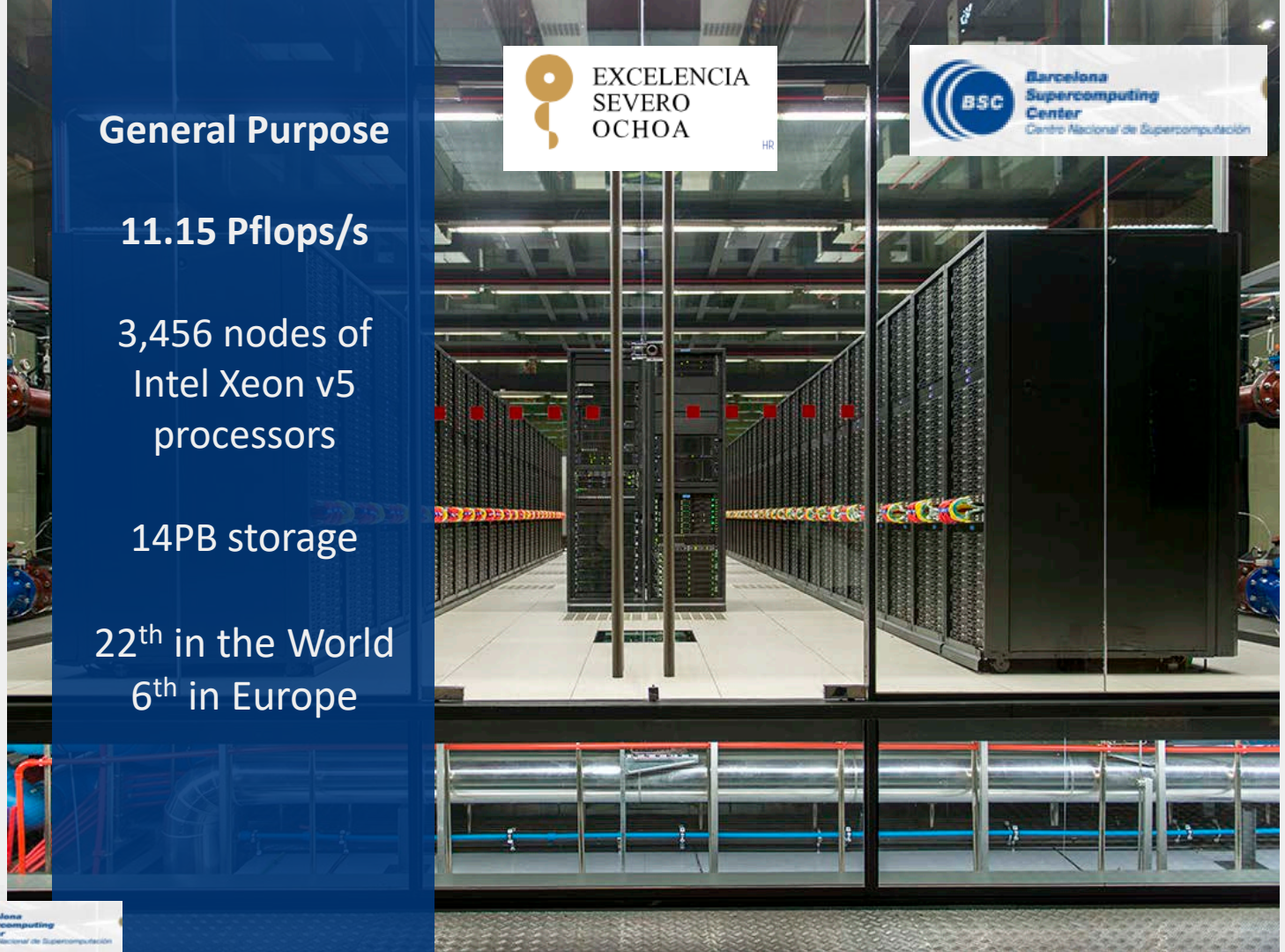
excelerate

elixir AAI

RD Connect

Transbionet network (INB hosted)





General Purpose

11.15 Pflops/s

3,456 nodes of Intel Xeon v5 processors

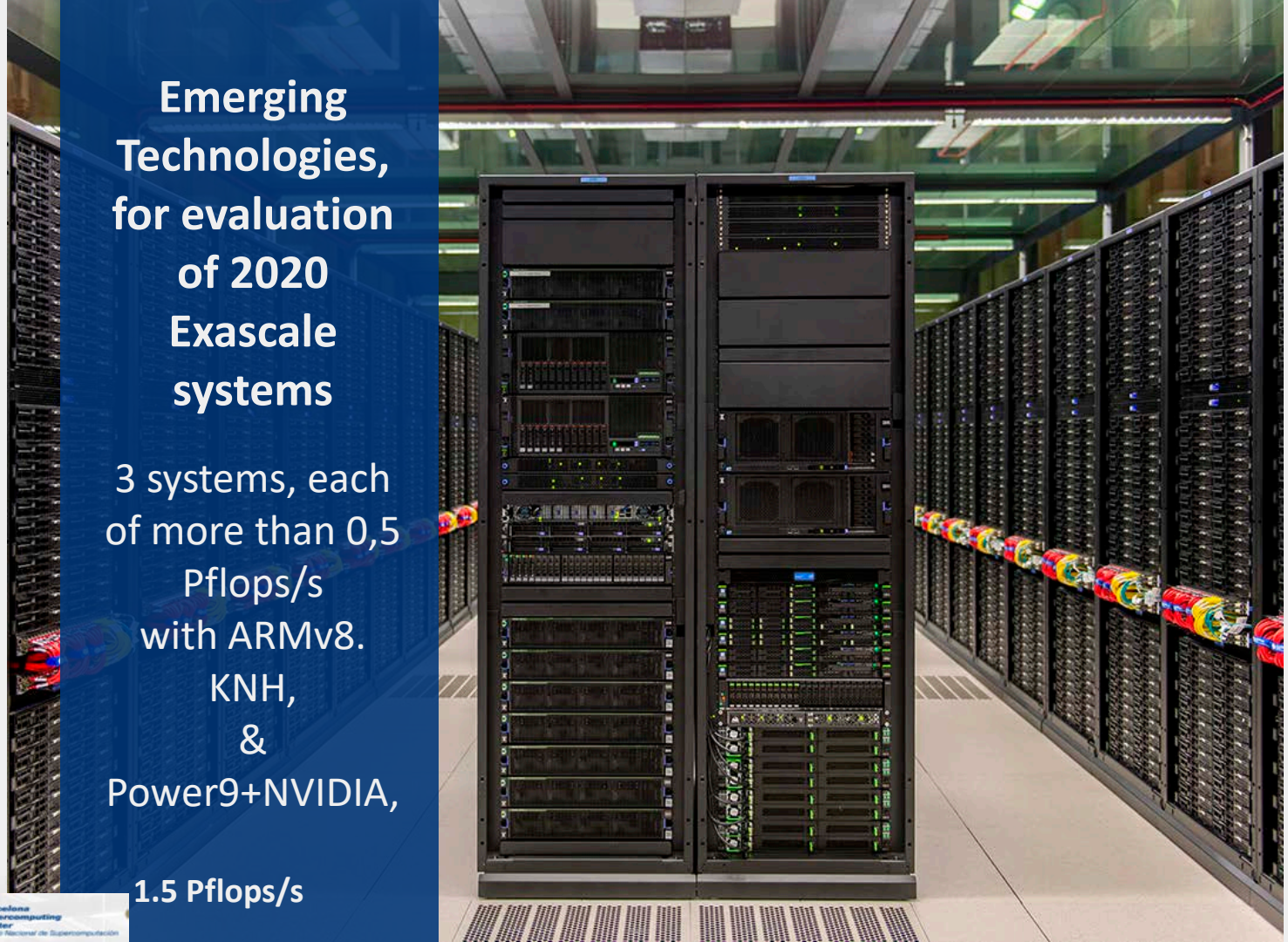
14PB storage

22th in the World
6th in Europe

EXCELENCIA SEVERO OCHOA

Barcelona Supercomputing Center
Centro Nacional de Supercomputación

If you want to know more, see…



Emerging Technologies, for evaluation of 2020 Exascale systems

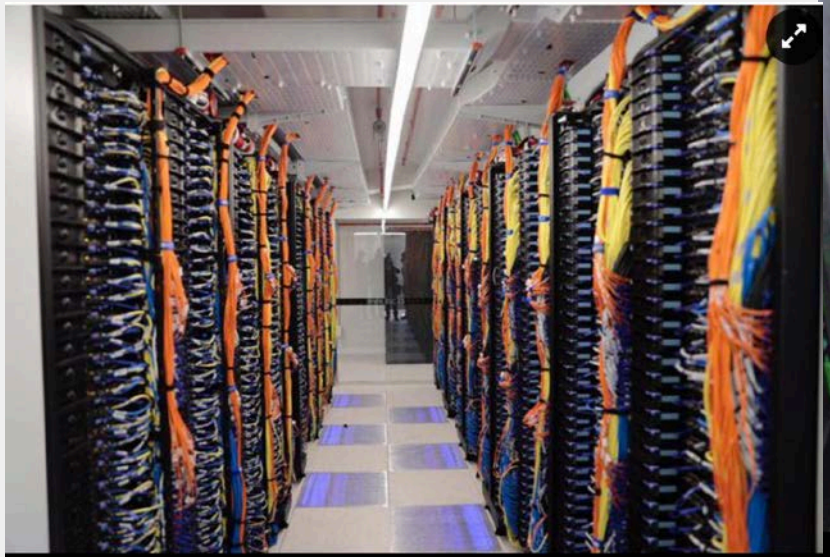
3 systems, each of more than 0,5 Pflops/s with ARMv8. KNH, & Power9+NVIDIA,

1.5 Pflops/s

Llega StarLife, un superordenador exclusivo para ciencias de la vida

• El Barcelona Supercomputing Center refuerza su apuesta estratégica por la investigación biomédica

LA VANGUARDIA



Se pone en marcha StarLife, una nueva infraestructura informática para impulsar la investigación biomédica (Xavier Cervera)

El sistema prestará servicio a investigadores de todo el mundo a través del Archivo Europeo de Genomas y Fenomas (Xavier Cervera)

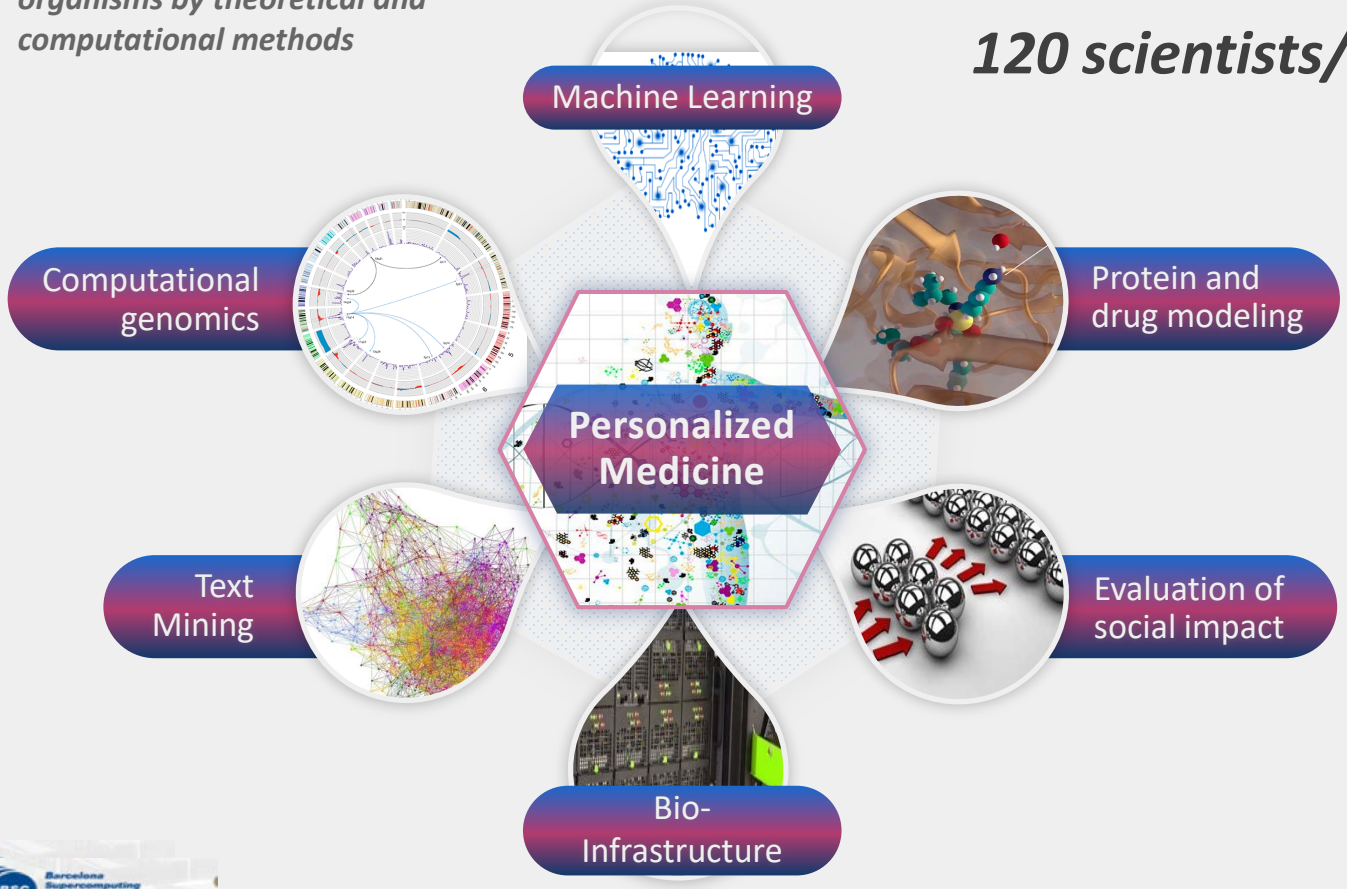
Life Sciences

Understanding living organisms by theoretical and computational methods

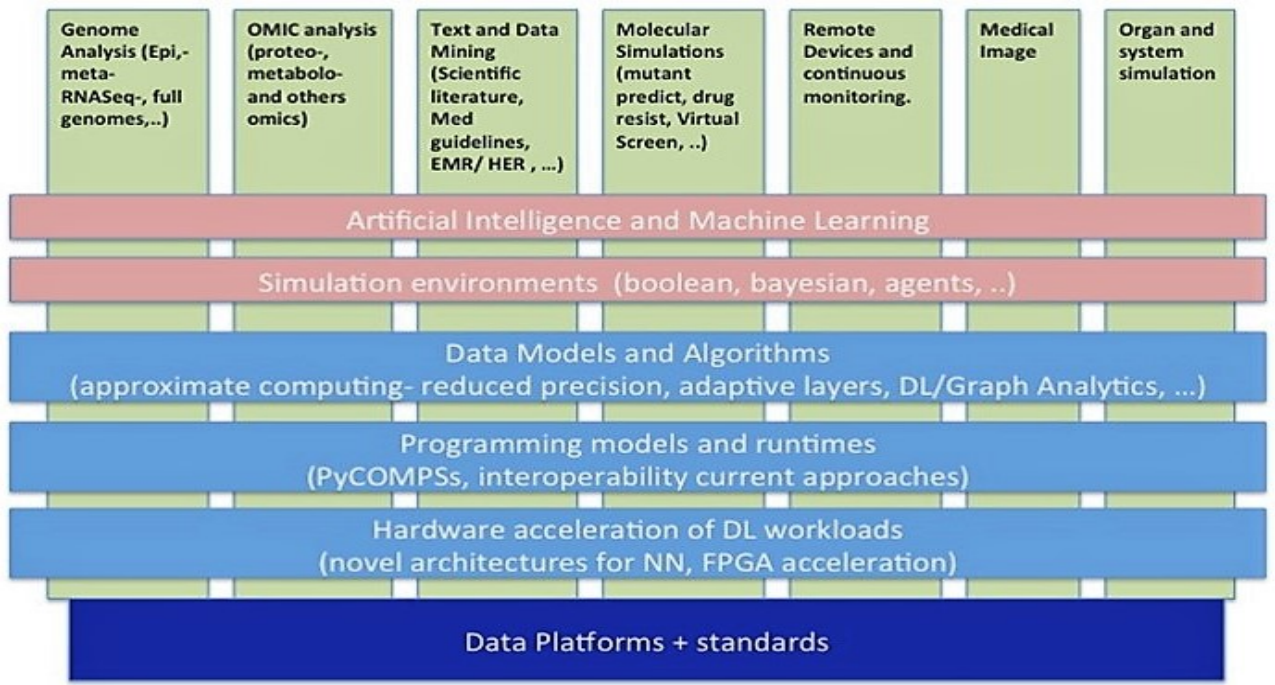
7 research groups

5 Support Units (including INB)

120 scientists/engineers by 2020



BSC technical strategy for Personalized Medicine



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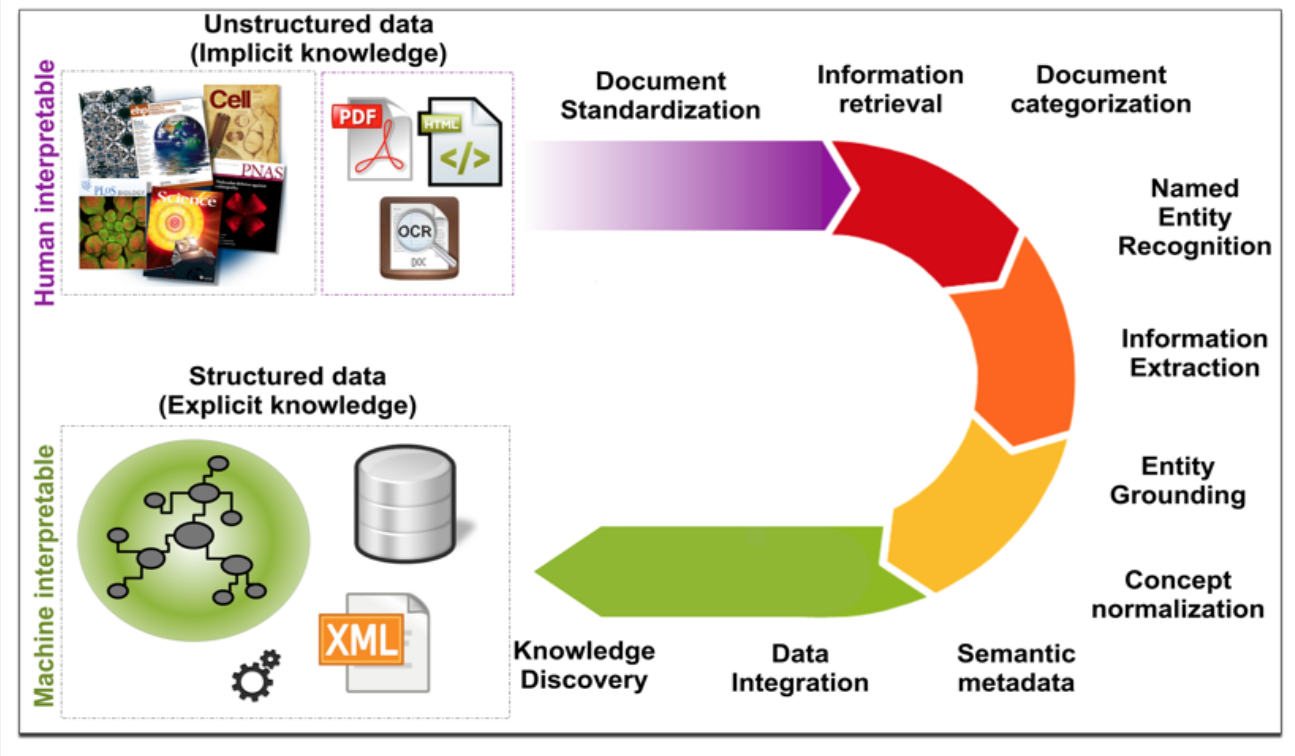
Practical utility in the clinic

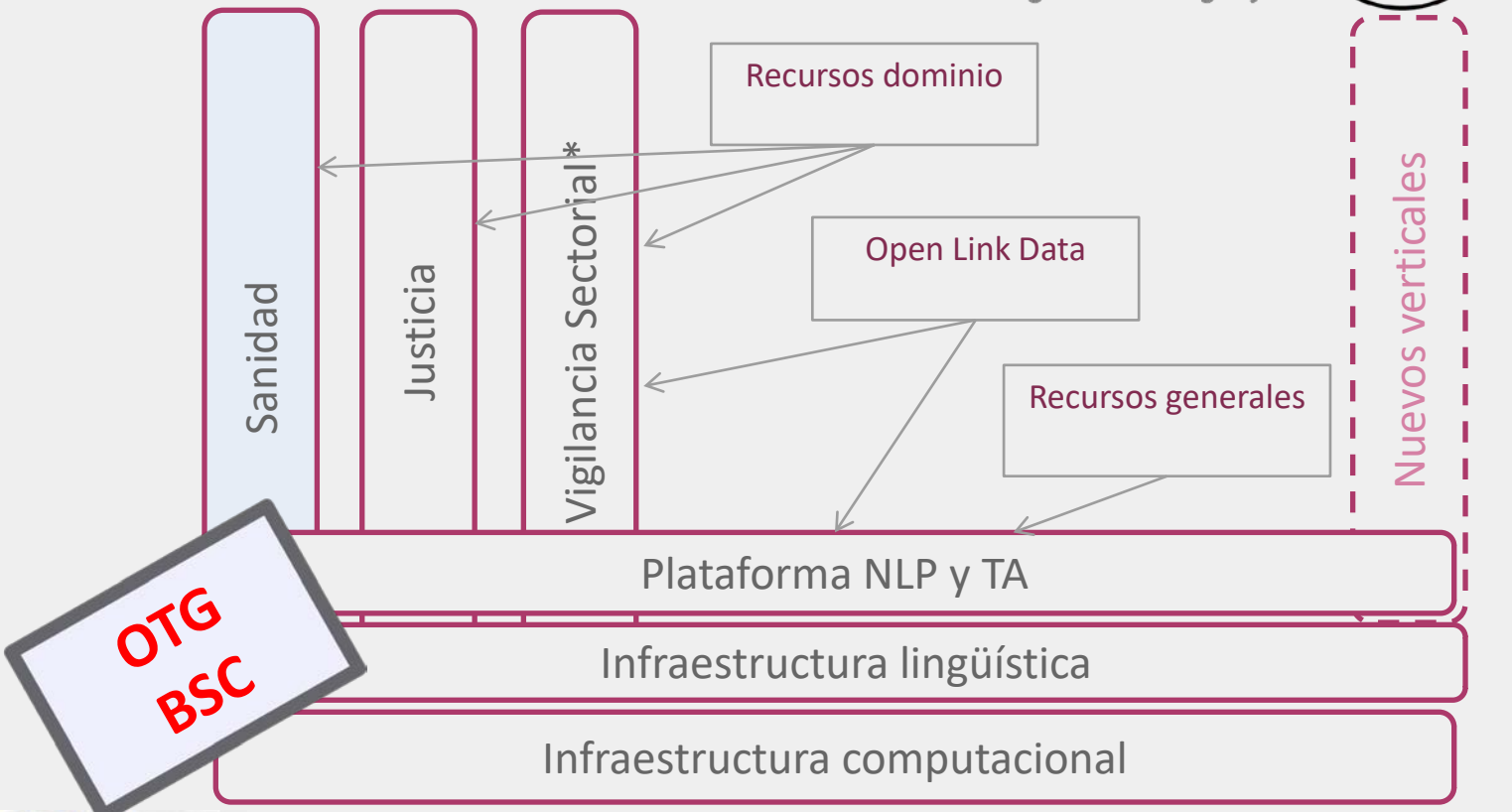
Mining EHR
Social Media
Medical Imaging
Other Health Data (papers, patents, etc)
Medical devices
Epidemiology

Gomez-Lopez (2017) Briefings in Bioinformatics,

NLP and Text Mining

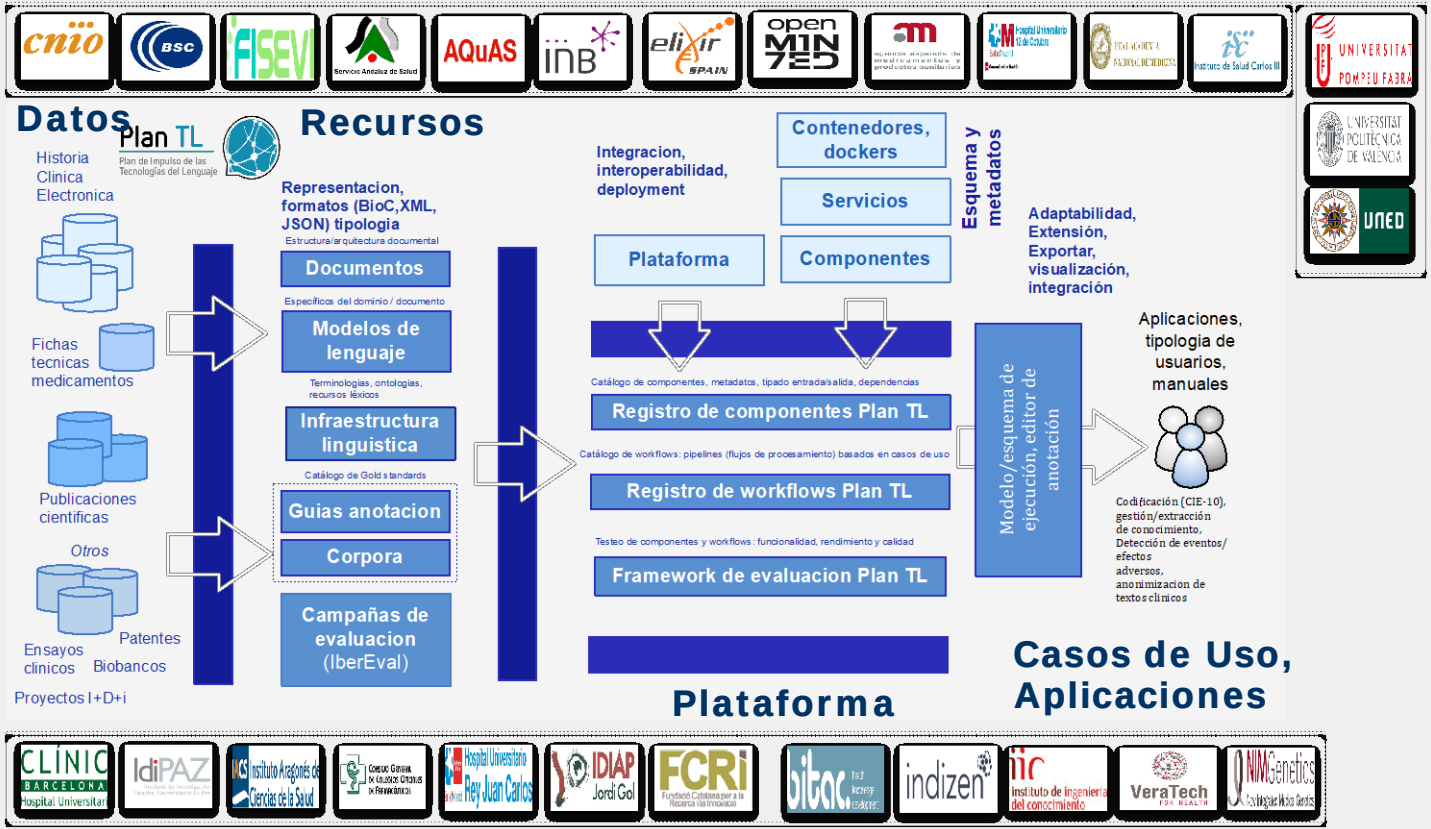


Plan TL
Plan de Impulso de las Tecnologías del Lenguaje



*Inteligencia competitiva

Plataforma TL componentes



nature physics

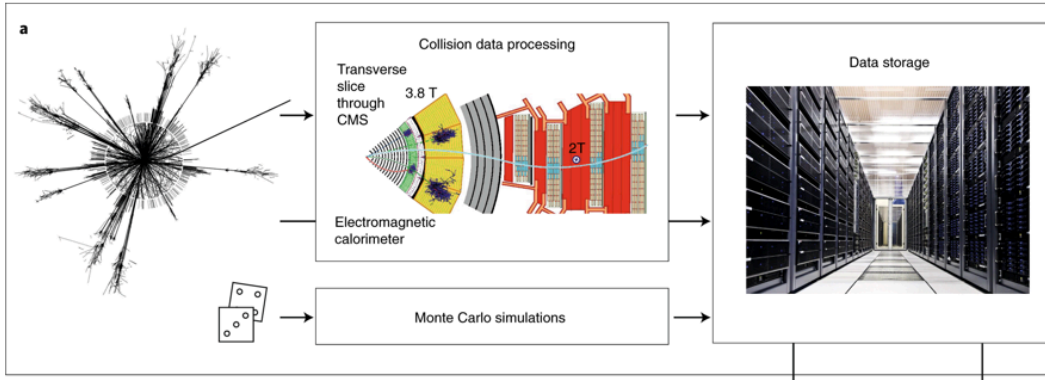
PERSPECTIVE

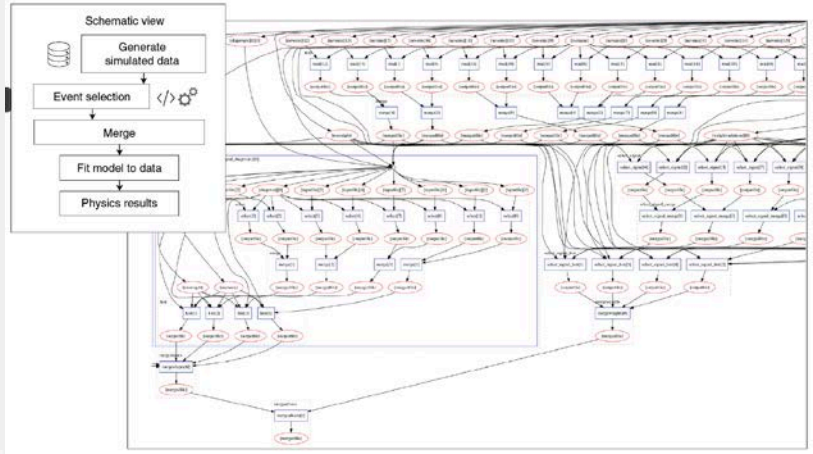
https://doi.org/10.1038/s41567-018-0342-2

Corrected: Publisher Correction

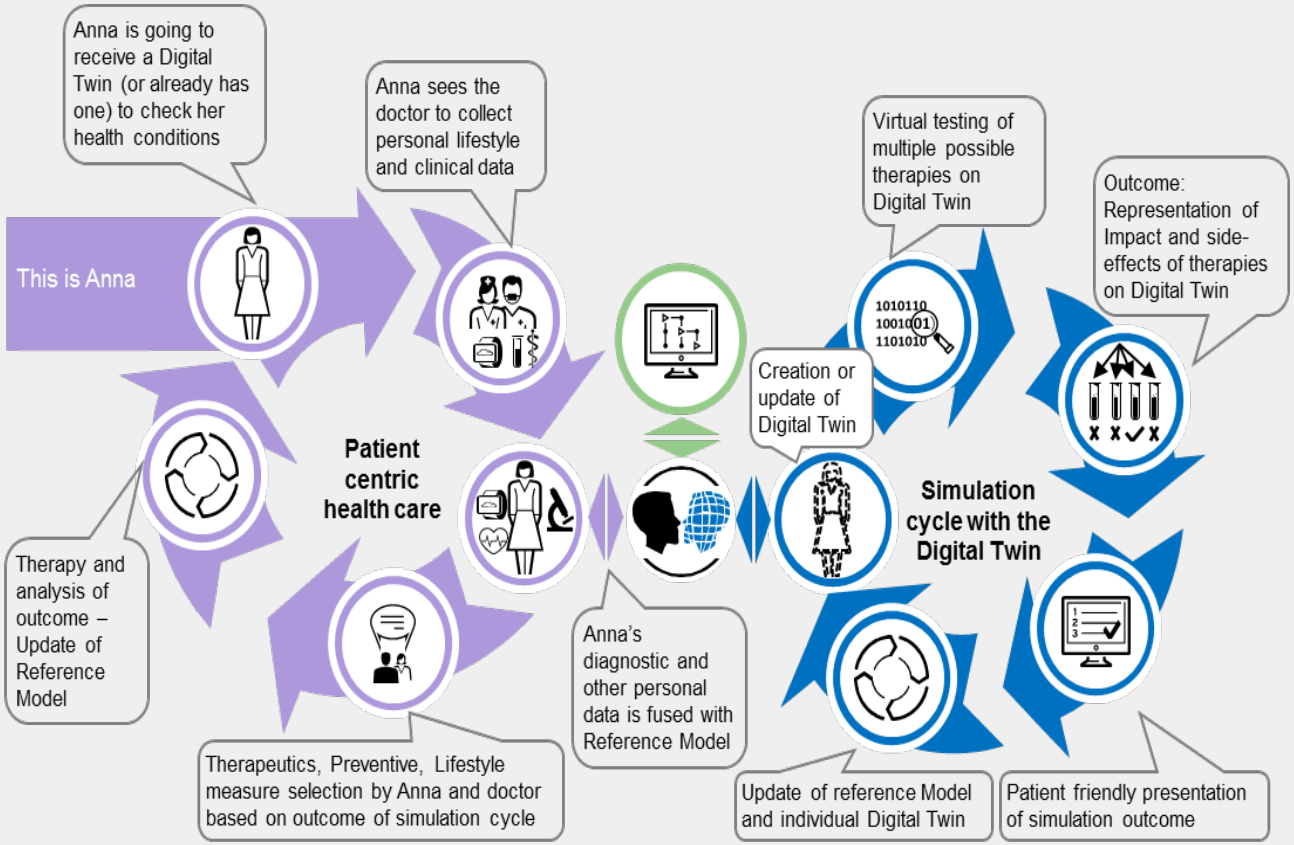
creenshot

Open is not enough





Digital Twins view of Future Medicine

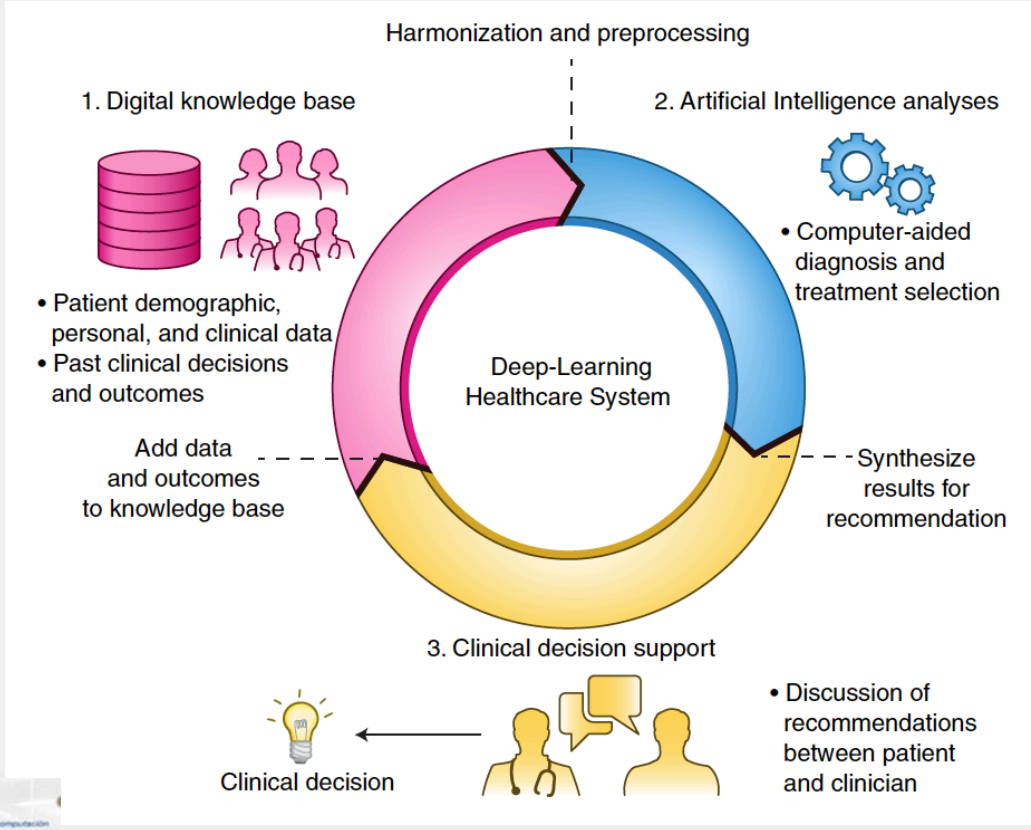


A call for deep-learning healthcare

Here we argue that now is the time to create smarter healthcare systems in which the best treatment decisions are computationally learned from electronic health record data by deep-learning methodologies.

Beau Norgeot, Benjamin S. Glicksberg and Atul J. Butte

NATURE MEDICINE | VOL 25 | JANUARY 2019 | 14–18 |



Nearly every other industry uses data on previous actions and outcomes to enable smarter ongoing choices. Amazon targets product recommendations to a user on the basis of similar customers' shopping patterns. Google updates its search results using the outcomes of previous searches as a basis. Waze uses information from drivers traveling similar routes to optimize the directions it provides. Why is medicine, as an industry, still left out?

It is time to safely bring huge medical data repositories and advanced learning algorithms together with physicians to make a deep-learning healthcare system. Deep learning, the newest iteration of machine-learning methodologies, is now performing at state-of-the-art levels in previously difficult tasks, including image analysis, language processing, information retrieval, and forecasting. Deep learning is well suited for medical data as it can identify patterns

birth.htm). But the US medical system is a competitive one, meaning that competing health systems, payers, and pharmaceutical companies are not incentivized to fully share clinical data, pricing of services or medications, and costs of the delivered care.

input-feature engineering. Current successes have shown performance that meets or

importantly, these systems can be run in real-time within or across entire hospital systems. We propose that future physicians will be armed with insights from models continuously trained and updated on real-world clinical data to make more accurate diagnoses and individually optimized

While EHRs have known challenges[1,2], they now represent a patient's legal medical record and are complete enough to enable a new physician to completely take over the care of a patient. This data is perhaps among the most expensive in the US, given that physicians are paid to enter much of it. Of course, EHR data must only be used in manners that are safe and respectful to the patients from which they are obtained, but it

panel of the very best physicians. Since deep-learning models could be shared between hospitals without the privacy risks of sharing patient data, there is nearly limitless potential to create a new system of precision medicine learned from the decisions and outcomes of diverse physicians treating diverse patients (Fig. 1).

A call for deep-learning healthcare

Here we argue that now is the time to create smarter healthcare systems in which the best treatment decisions are computationally learned from electronic health record data by deep-learning methodologies.

Beau Norgeot, Benjamin S. Glicksberg and Atul J. Butte

Two incredible information technology revolutions are currently colliding in medicine. Electronic health records (EHRs) are capturing the thoughts and orders of the best-trained physicians and images and outcomes from their treated patients, and advances in machine learning are beginning to supplement clinical medicine. But breakthroughs still remain fully unrealized because these revolutions are siloed. While there are raw materials from which current actions and outcomes in medicine could be learned, they are still not systematically utilized to improve the practice of medicine.

Nearly every other industry uses data on previous actions and outcomes to enable smarter ongoing choices. Amazon targets product recommendations to a user on the basis of similar customers' shopping patterns. Google updates its search results using the outcomes of previous searches as a basis. Waze uses information from drivers traveling similar routes to optimize the directions it provides. Why is medicine, as an industry, still left out?

The roadblocks to bringing medicine into the data-driven era are operational and cultural. Many have written about inefficiencies in the US medical system in regard to rising healthcare spending (http://apps.who.int/gho/data/node.main.GHEDCHEGDPSHA2011) and the challenges in improving quality (https://data.oecd.org/healthstat/life-expectancy-at-birth.htm). But the US medical system is a competitive one, meaning that competing health systems, payers, and pharmaceutical companies are not incentivized to fully share clinical data, pricing of services or medications, and costs of the delivered care. There is a significant room for improvement and potential to better use the data that are available.

While EHRs have known challenges[1,2], they now represent a patient's legal medical record and are complete enough to enable a new physician to completely take over the care of a patient. This data is perhaps among the most expensive in the US, given that physicians are paid to enter much of it. Of course, EHR data must only be used in manners that are safe and respectful to the patients from which they are obtained, but it will be a significant regret if this data is not used to improve the practice of medicine.

Over 10 years ago, Lynn Etheredge[3] and others[4] proposed the Learning Health System, whereby millions of EHRs could be used to inform medical practice and policy. But these visionaries were proposing a system in which physicians mediate the learning. Now with nearly 80% of medication orders captured electronically, more than 1.7 billion prescriptions per year electronically tracked[5], and 98% of hospital systems using EHRs (https://dashboard.healthit.gov/quickstats/pages/FIG-Hospitals-EHR-Incentive-Programs.php), we can envision computer systems that learn how to improve the medical system by themselves.

It is time to safely bring huge medical data repositories and advanced learning algorithms together with physicians to make a deep-learning healthcare system. Deep learning, the newest iteration of machine-learning methodologies, is now performing at state-of-the-art levels in previously difficult tasks, including image analysis, language processing, information retrieval, and forecasting. Deep learning is well suited for medical data as it can identify patterns in sparse, noisy data and requires little input-feature engineering. Current successes have shown performance that meets or surpasses that of experts, but perhaps more importantly, these systems can be run in real-time within or across entire hospital systems. We propose that future physicians will be armed with insights from models continuously trained and updated on real-world clinical data to make more accurate diagnoses and individually optimized treatment decisions.

Is there one optimal way to practice medicine? Imagine ten physicians faced with a single clinical conundrum (choice A, B, or C) in one patient. If these ten were provided with the maximum possible amount of information about a patient in a clear format, from physical exam results to wearables to the patient's own preferences for care, the world's biomedical literature and abstracts, and data on similar patients, in any desired format, should all ten physicians reach the exact same choice for this clinical decision? We know today that they probably would not, but should they? If the answer is yes, then medicine is fundamentally machine learnable.

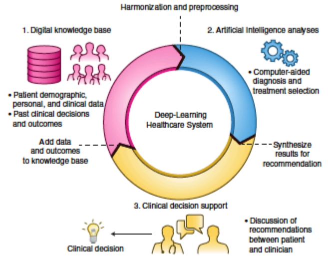


Fig. 1 | **A deep-learning healthcare system.** A schematic representation of a deep-learning healthcare system is shown.

Many health conditions present in heterogeneous ways, making it challenging to establish an accurate diagnosis over time. The treatment regimens for many complex conditions require physicians to stay aware of the latest options and evidence. A deep-learning healthcare system would enable all physicians to practice at the same level of expertise as a panel of the very best physicians. Since deep-learning models could be shared between hospitals without the privacy risks of sharing patient data, there is nearly limitless potential to create a new system of precision medicine learned from the decisions and outcomes of diverse physicians treating diverse patients (Fig. 1).

Fogel and Kvedar[6] have insightfully noted that bringing artificial intelligence to medicine will not sideline doctors, but will instead enhance their strengths. Physicians, empowered by patterns and evidence derived from large-scale real-world practice data, will be able to focus on the uniquely human elements of their profession for which they are best trained. Tasks that cannot be performed by a machine because they require emotional intelligence, such as asking the patient careful questions to uncover more nuanced symptoms and building trust through personal relationships by using human intuition, will still be unique qualifications of physicians to guide the implementation of the computationally optimized diagnoses and treatment plans of the future. ❐

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Competing interests

A.J.B. reports grants and non-financial support from Progenity, personal fees from NuMedii, personal fees from Personalis, grants and personal fees from NIH (multiple institutes), grants from L'Oreal, grants and personal fees from Genentech, personal fees from Merck, personal fees from Lilly, personal fees from Assay Depot, personal fees from Geisinger Health, personal fees from GNS Healthcare, personal fees from uBiome, personal fees from Roche, personal fees from Wilson Sonsini Goodrich & Rosati, personal fees from Orrick, Herrington & Sutcliffe, personal fees from Verinata, personal fees from 10x Genomics, personal fees from Pathway Genomics, personal fees from Guardant Health, personal fees from Gerson Lehrman Group, personal fees from Nuna Health, personal fees from Samsung, personal fees from Milken Institute, personal fees from Brown University, personal fees from Oregon Health Sciences University Knight Cancer Center, personal fees from Vermont Oxford Network, personal fees from University of Chicago, personal fees from Mount Sinai School of Medicine, personal fees from University of Pittsburgh School of Medicine, personal fees from Capital Royalty Group, personal fees from Champalimaud Foundation, personal fees from Scripps Translational Science Institute, personal fees from Washington University in St. Louis, personal fees from University of Maryland, personal fees from HIMSS, personal fees from Federation of Clinical Immunology Societies (FOCIS), personal fees from Kansas City Area Life Sciences Institute, personal fees from Association for Molecular Pathology, personal fees from TEDMED, personal fees from Physician's Education Resource, personal fees from Optum Labs, personal fees from National Jewish Health, personal fees from Federation of the Israeli Societies for Experimental Biology (FISEB), personal fees from Pfizer, personal fees from Bayer, personal fees from Fusion Conferences, personal fees from Accelerating Biopharmaceutical Development (AccBio), personal fees from Three Lakes Partners, personal fees from Pediatric Academic Societies, personal fees from Korean Society for Biochemistry and Molecular Biology, personal fees from Human Proteomics Organization (HUPO), personal fees from HudsonAlpha, personal fees from Tensegrity, grants from Intervalien Foundation, personal fees from Association for Academic Health Sciences Libraries, personal fees from Westat, personal fees from FH Foundation, personal fees from University of Kentucky, personal fees from University of Pennsylvania, personal fees from The Transplantation Society, personal fees from California Office of Planning and Research, personal fees from WuXi, personal fees from University of Arkansas, personal fees from FlareCapital, personal fees from National Academies, personal fees from Helix, personal fees from American Urological Association, personal fees from Association for American Medical Colleges, personal fees from Roam Insights, personal fees from United Network for Organ Sharing, personal fees from American Association of Allergy Asthma and Immunology, personal fees from University of Michigan, personal fees from Autodesk, personal fees from Regenstrief Institute, personal fees from American Medical Association, personal fees from Precision Medicine World Conference, personal fees from University of Chicago, personal fees from Mars, and personal fees from Kneed Media. 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US20170224683." pending, and Stanford University pays royalties each year on licensed intellectual property. MIT Press pays royalties each year on book sales.

Cardiologist-level arrhythmia detection and classification in ambulatory electrocardiograms using a deep neural network

Awni Y. Hannun[1,6]*, Pranav Rajpurkar[1,6], Masoumeh Haghpanahi[2,6], Geoffrey H. Tison[3,6], Codie Bourn[2], Mintu P. Turakhia[4,5] and Andrew Y. Ng[1]

nature medicine

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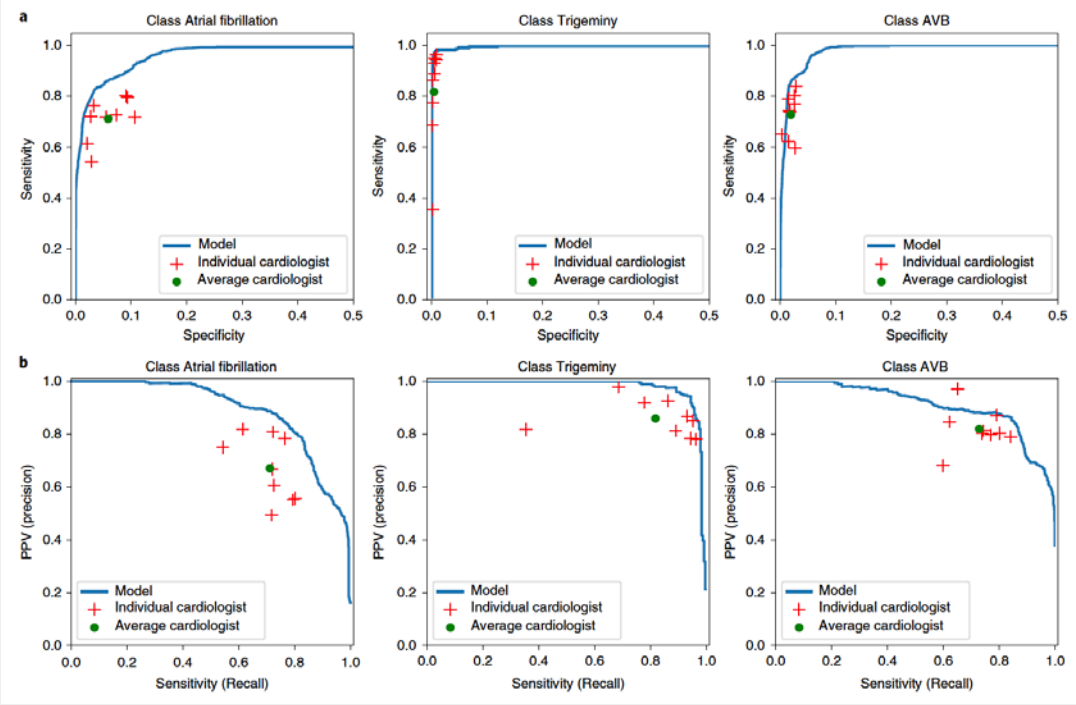

IMI Lesson I am learning (1/2)

Symmetry of the problems between companies and academia

- information silos in departments or administrations
- difficult to adhere to long term projects
- limited capacity to adapt to new technologies

Data Management is Key

- Federation for searches and execution (beacons and containers)
- Encryption (i.e. searches in encrypted data)
- Trusted partners as mediators
- FAIR / standards to facilitate the internal operations

Thrust (has a lot to do with exploration versus stability)

IMI Lesson I am learning (2/2)

New calls

- Data management issues
- Analysis platforms integrating OMICS and EHR (others)
  (there are many call - projects on-going)
- AI/ML applications on diverse data sets and applications

New projects should be more challenging and transformative.

XII CONFERENCIA ANUAL
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Darío Garcia-Gasulla, Ferran Pares, Armand Vilalta BSC (High Performance AI)

Roland Mathis, Matteo Manica IBM (Computational systems biology)

Vera Pancaldi CRCT, Toulouse

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Spanish MINECO
"Comorbidity networks"

BLUEPRINT epigenome

eTOX

eTRANSAFE

efpia imi

OPENMINDTED

BioCreAtIvE

Excellerate-ELIXIR

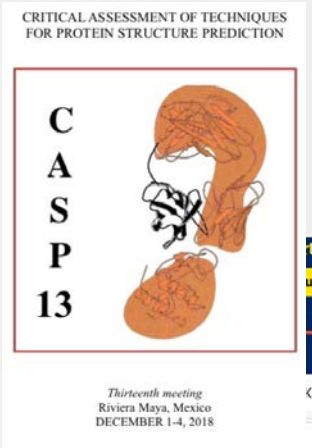
elixir SPAIN

INB Instituto Nacional de Bioinformática

ISCB International Society for Computational Biology

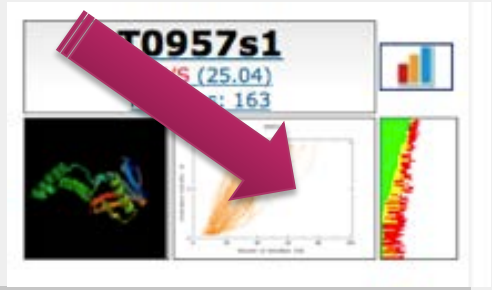
fellows

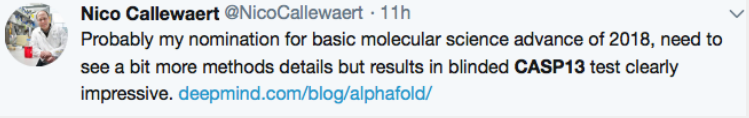
Alfons_Valencia



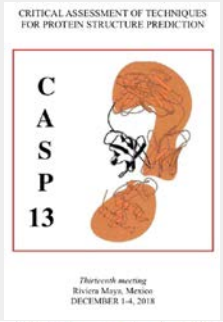
Predicting protein structure from the sequence is one of the fundamental problems in molecular biology.

It is the key to the prediction of the consequences of mutations in human diseases and to drug design

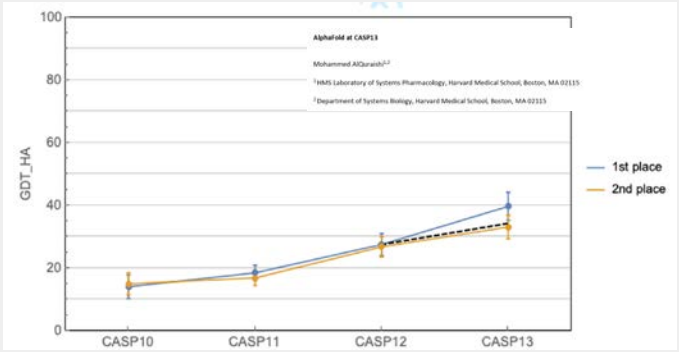


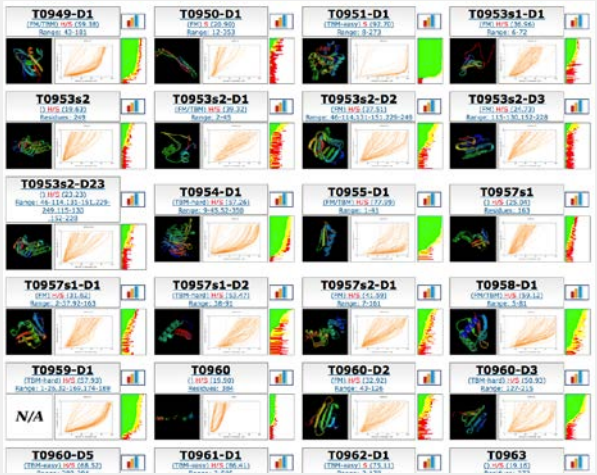


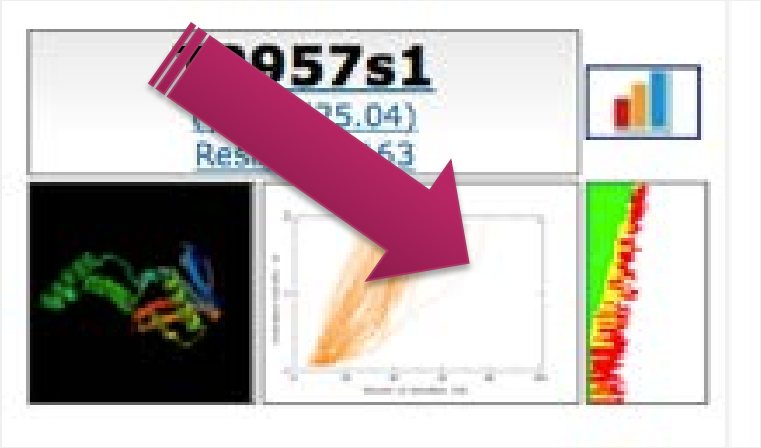
On its first foray into the competition, AlphaFold topped a table of 98 entrants, predicting the most accurate structure for 25 out of 43 proteins, compared with three out of 43 for the second placed team in the same category.



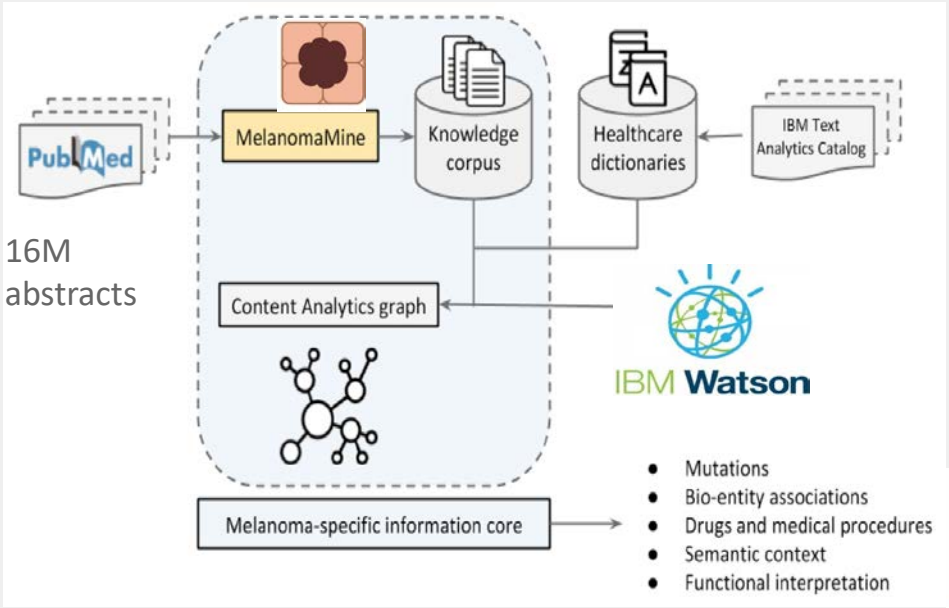
All CASP results are available (each target / all predictors)







Data mining: **Complex Networks**

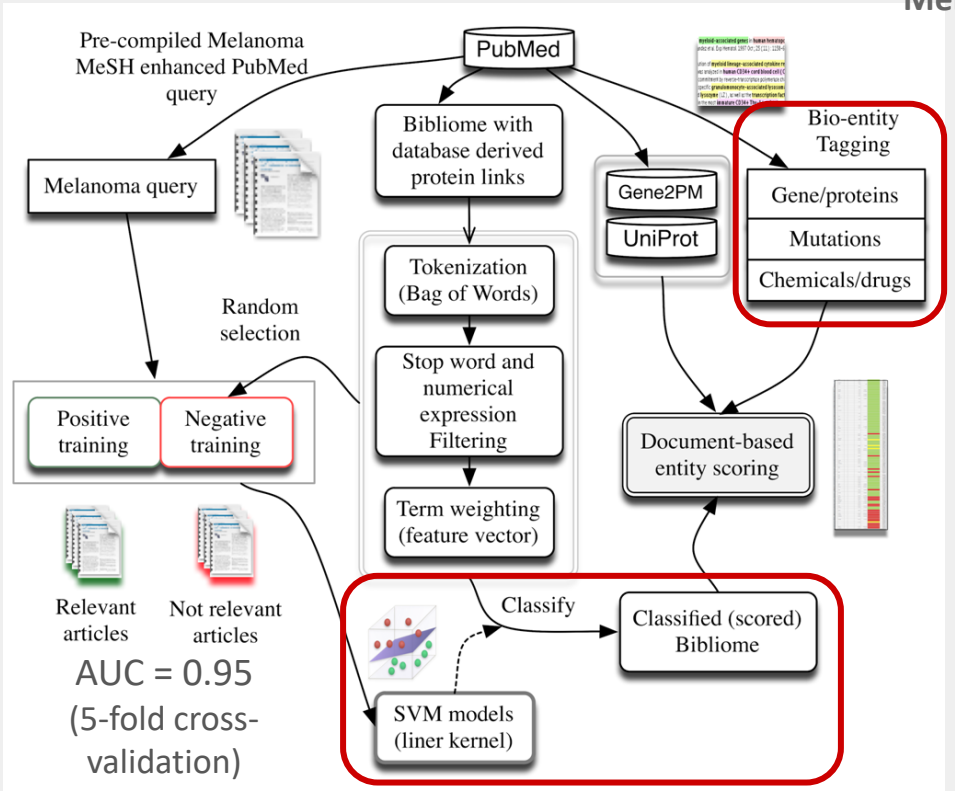


Melanoma content analytics with MelanomaMine and IBM Watson

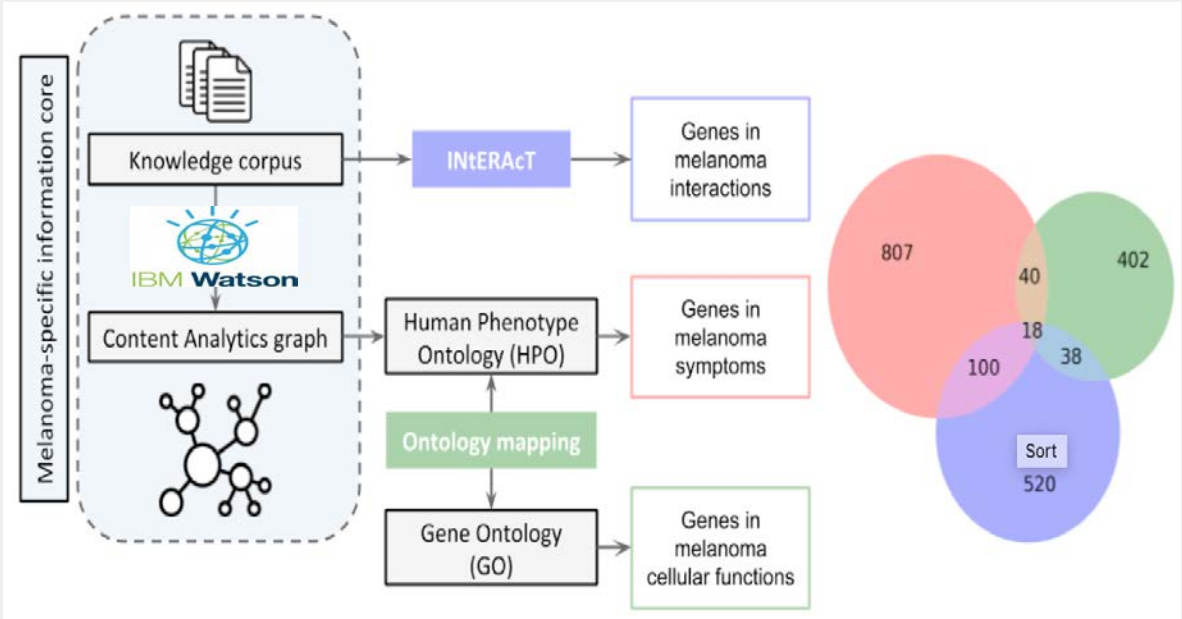
Davide C[1], Andres E[2], Corvi J[3], Fernandez Gonzalez JM[3], Lopez-Martin JA[4], Capella-Gutierrez S[3], Krallinger M[5], Valencia A[1,6] **Submitted 2019**

- Bio-entities associations
- Functional interpretation
- Translational applications

MelanomaMine



Data mining: **Complex Networks**



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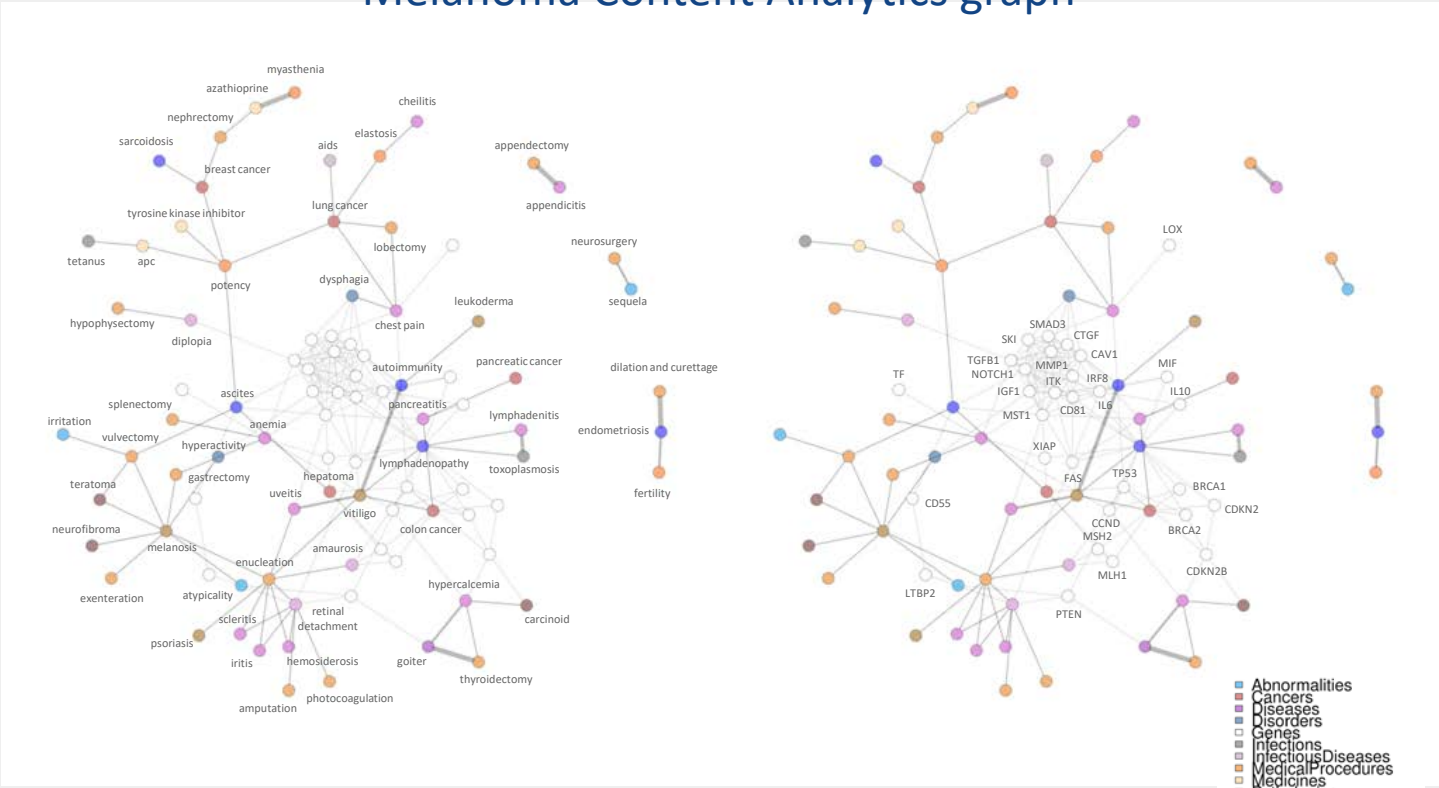
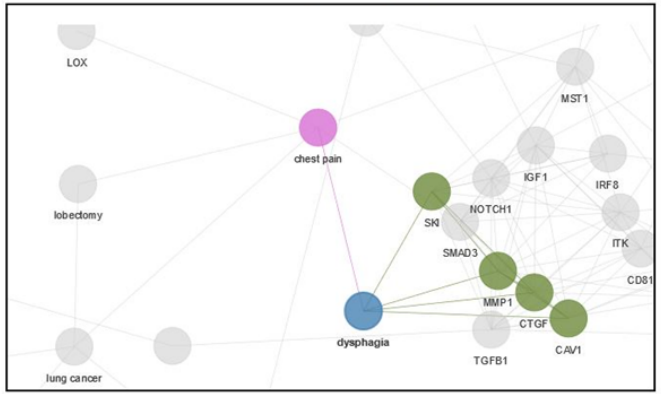
Melanoma Content Analytics graph



Data mining: **Complex Networks**

Melanoma Content Analytics graph



id	group	Select
lung cancer	Cancers	☐
chest pain	Symptoms	☑
dysphagia	Disorders	☑

Selected:

chest pain,dysphagia

PMID	sentence
1916958	He presented with DYSPHAGIA and weight loss.
2271301	We present one case of primary malignant melanoma of the esophagus in a 76-year-old woman who reported the symptoms of DYSPHAGIA and recent weight loss; the radiography showed a large polypoid mass filling the entire lower half of the esophagus, dark brown-black in the endoscopy.
3792282	We report the case of a 74-year-old woman who was admitted to hospital with progressive DYSPHAGIA.
3964453	It may cause significant local symptoms such as airway obstruction and DYSPHAGIA, and, in some cases, may represent the initial manifestation of disseminated disease.
8493638	A 75 year old man with an eight month history of DYSPHAGIA and weight loss underwent pericardiocentesis for a massive pericardial effusion.
9369967	A 48-year-old woman presented with a 6-month history of DYSPHAGIA, often associated with retrosternal CHEST PAIN.
18773432	The patient is a 76-year-old male, who presented in the emergency room with bilateral CHEST PAIN exacerbated by inspiration.
18826613	CASE PRESENTATION: A progressive severe DYSPHAGIA case is reported induced by a melanoma of unknown origin (metastatic to a posterior mediastinal lymph node). [...] INTRODUCTION: We describe an original case of progressive severe DYSPHAGIA caused by a posterior mediastinal metastatic melanoma of unknown origin.
19212291	CASE REPORT: We report an original observation of an 82-year-old man with a pulmonary nodule presenting with CHEST PAIN.

R SHINY APP

Improving the interpretation of standard pathway analysis by linking ontological annotations